Determining Socioeconomic Impact of Rare Disease

Canadian Organization for Rare Disorders
Fall Conference 2023, Calgary, AB

Canada's Rare Disease Landscape Creating Knowledge and Cures, 3:15pm – 4:55pm

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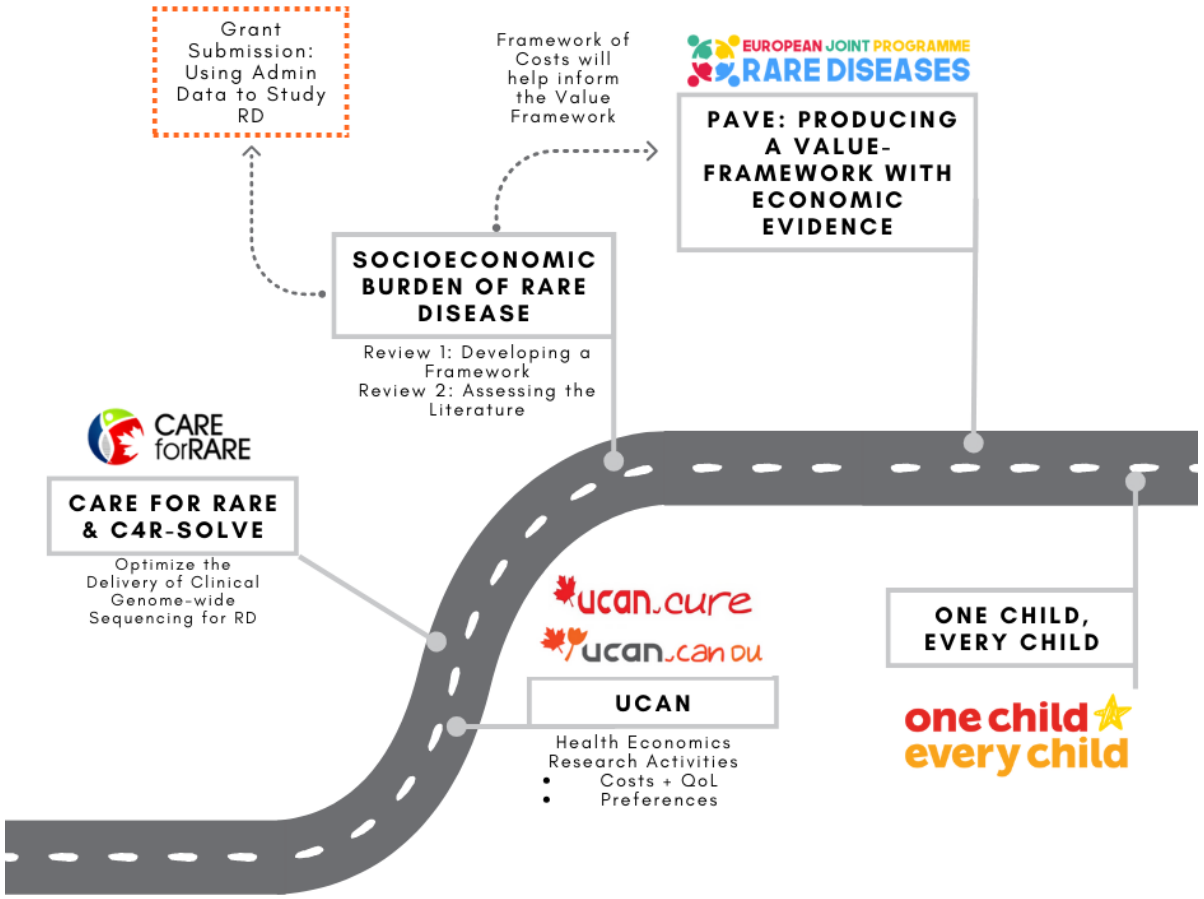
Deborah Marshall, PhD
University of Calgary

November 29, 2023

Research Program Overview:

Building on our understanding of the socioeconomic burden of RD



CIHR IRSC Canadian Institutes of Health Research / Instituts de recherche en santé du Canada
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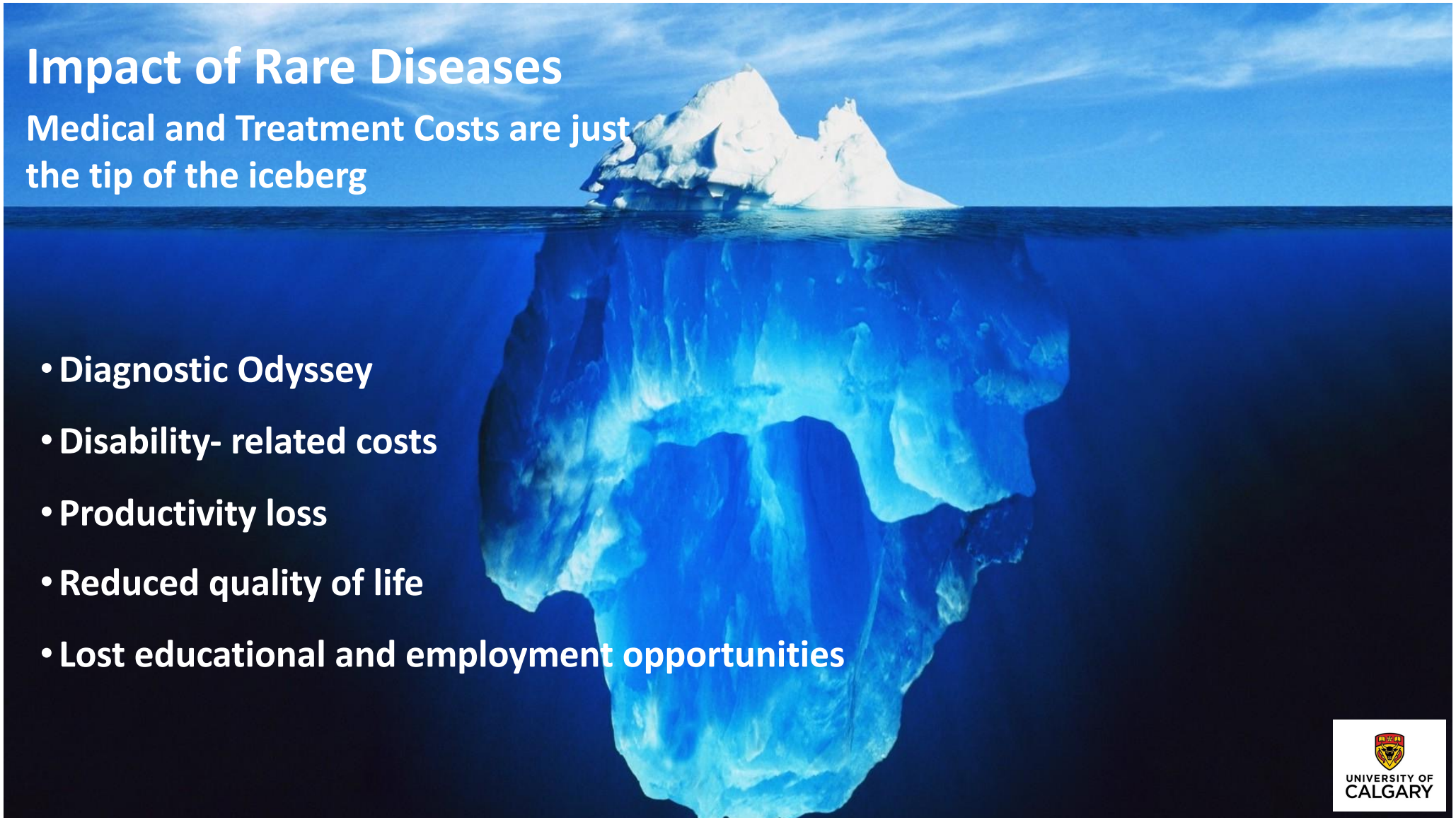
EUROPEAN JOINT PROGRAMME RARE DISEASES

Impact of Rare Diseases

Medical and Treatment Costs are just the tip of the iceberg



- Diagnostic Odyssey
- Disability- related costs
- Productivity loss
- Reduced quality of life
- Lost educational and employment opportunities

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Costs of RD: Developing a Framework of Costs

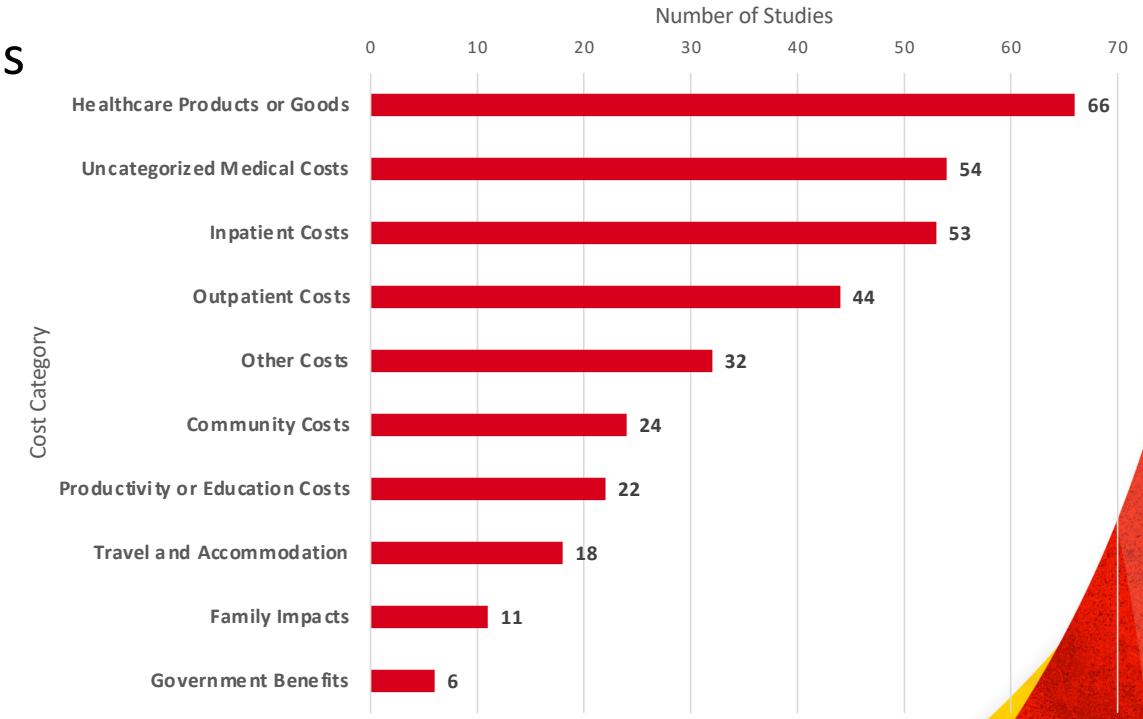
We identified existing frameworks for measuring cost elements of SEB for chronic or rare diseases + drew on expert input to develop a standardized list of cost elements

- **Inpatient costs** (e.g., hospitalization)
- **Outpatient costs** (e.g., emergency room visits)
- **Community costs** (e.g., paid care)
- **Healthcare products or goods** (e.g., prescriptions)
- **Productivity or education costs** (e.g., missed school or work)
- **Travel and accommodation** (e.g., transportation)
- **Government benefits** (e.g., disability)
- **Family impacts** (e.g., adaptations)
- **Other costs relevant to rare disease** (e.g., out of country travel for advanced testing or treatment)

Currie, G. R., Gerber, B., Lorenzetti, D., MacDonald, K., Benseler, S. M., Bernier, F. P., Boycott, K. M., Carias, K. V., Hamelin, B., Hayeems, R. Z., LeBlanc, C., Twilt, M., van Rooijen, G., Wong-Rieger, D., Yeung, R. S. M., & Marshall, D. A. (2023). Developing a Framework of Cost Elements of Socioeconomic Burden of Rare Disease: A Scoping Review. *PharmacoEconomics*, *41*(7), 803–818. https://doi.org/10.1007/s40273-023-01262-x

Costs of RD: What we found in the literature

- Most reported costs categories were health system focussed
- Less than half of the studies examined included:
  - Community Costs
  - Productivity or Education Costs
  - Travel and Accommodation
  - Family Impacts
  - Government Benefits



Marshall, D. A., Gerber, B., Lorenzetti, D. L., MacDonald, K. V., Bohach, R. J., & Currie, G. R. (2023). Are We Capturing the Socioeconomic Burden of Rare Genetic Disease? A Scoping Review of Economic Evaluations and Cost-of-Illness Studies. *PharmacoEconomics*, *41*(12), 1563–1588.

The Diagnostic Odyssey

- Our studies have shown a lengthy diagnostic odyssey among patients with suspected genetic rare disease, taking place over several years, and requiring multiple diagnostic-related tests
  - Average of **5.5 years** and **16 diagnostic tests** between symptom onset and WES (sub-sample of 228 patients enrolled in the SOLVE cohort, from Alberta and Ontario) [1]
  - Average of **3.2 years** and **24 diagnostic tests** between first diagnostic tests and WES (sub-sample of 369 patients enrolled in the SOLVE cohort, from Alberta and Ontario) [2]

[1] *Hayeems R, Michaels Igbokwe C, Venkataramanan V, Hartley T, Acker M, Gillespie M, Ungar W, Mendoza-Londona R, Bernier FP, Boycott K, Marshall DA. The complexity of diagnosing rare disease: An organizing framework for outcomes research and health economics based on real-world evidence. Genet Med 2022; 24(3), 694–702*

[2] *Seeger TA, Hayeems RZ, Hartley T, Venkataramanan V, Fooks K, MacDonald KV, Boycott KM, Bernier FP, Mendoza-Londona R, Ungar WJ, Marshall DA. Diagnostic Testing and Cost Pathways for Patients With Undiagnosed Suspected Rare Genetic Diseases (RGD): C4R Solve Multi-Centre Prospective Observational Cohort. CAHSPR Annual Conference 2023; Montreal, Quebec, Canada: May 29-31, 2023 (poster presentation).*

Substantial Out of Pocket Costs for Families of Children with Rare Disease (RD)

- 84% reported paying out of pocket for **costs related to obtaining a diagnosis**
- 83% reported paying out of pocket for **costs related to treatment**

Out of Pocket Costs Related to Diagnosis

Cost range	Frequency	Percent
$0	35	11.0%
$1 - $999	140	43.9%
$1000 - $4,999	73	22.9%
$5,000 - $49,999	47	14.7%
$50,000 - $99,999	4	1.3%
$100,000 or more	3	0.9%
Unsure	17	5.3%
Total	**319**	**100%**

Out of Pocket Costs Related to Treatment

Cost range	Frequency	Percent
$0	29	9.1%
$1 - $999	60	18.8%
$1000 - $4,999	70	21.9%
$5,000 - $49,999	107	33.5%
$50,000 - $99,999	11	3.4%
$100,000 or more	15	4.7%
Unsure	27	8.5%
Total	**319**	**100%**

Please do not cite or quote – UNPUBLISHED research in progress by Marshall et al

Direct Healthcare Costs for Children with RD

- The highest annual mean total cost for all cohorts occurred in the year after diagnosis (CAD$):

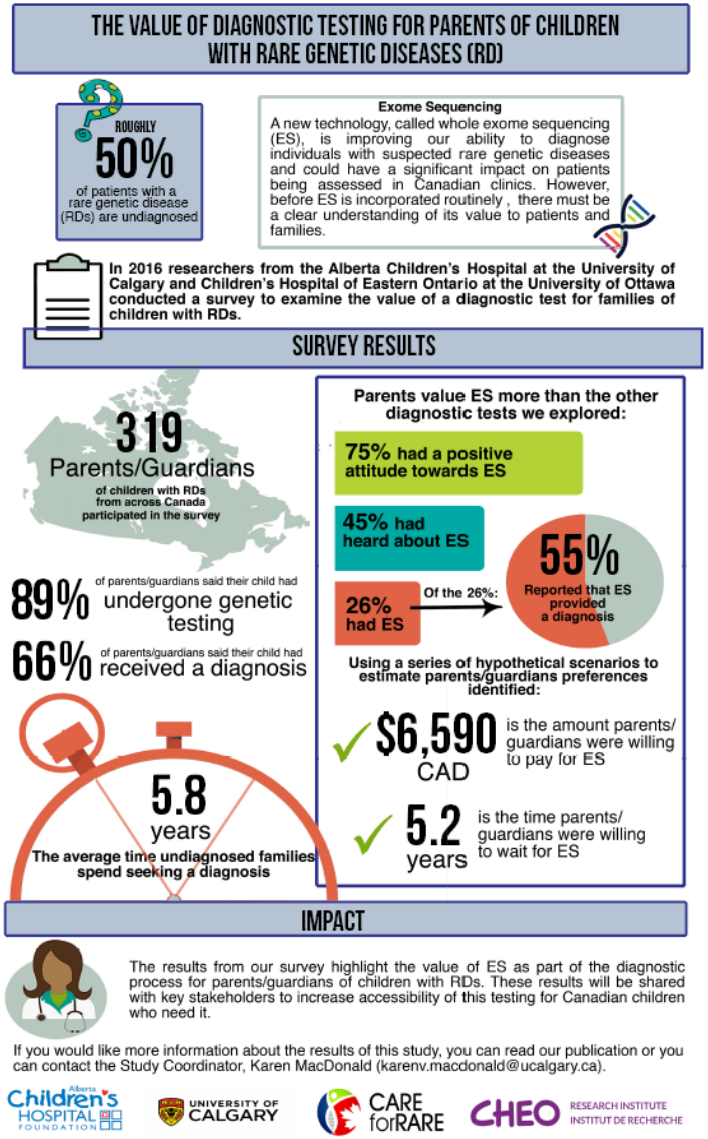
Rare Disease	Chronic Disease - Asthma	Chronic Disease - Diabetes	General Population
$23,333	$3,316	$4,890	$1,181

- During the post-diagnostic period, annual mean total costs for the genetic disease cohort were significantly higher than all other cohorts

- Marshall DA, Benchimol El, MacKenzie A, Rodriguez Duque D, MacDonald KV, Hartley T, Howley T, Hamilton A, Gillespie M, Malam F, Boycott KM. Direct health-care costs for children diagnosed with genetic diseases are significantly higher than for children with other chronic diseases. Genet Med. 2019;21(5):1049-1057

The Value of Knowing

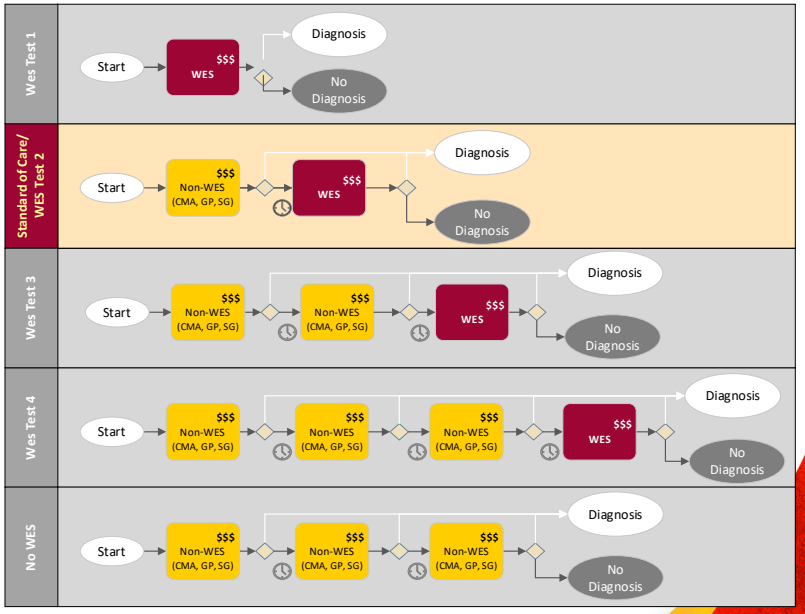
- Timely access to exome sequencing is highly valued by parents of children with rare diseases
- Parents were willing to pay CAD $6,590 for exome sequencing compared to other procedures to reduce the diagnostic odyssey



Marshall DA, Benchimol EI, MacKenzie A, Rodriguez Duque D, MacDonald KV, Hartley T, Howley T, Hamilton A, Gillespie M, Malam F, Boycott KM. Direct health-care costs for children diagnosed with genetic diseases are significantly higher than for children with other chronic diseases. Genet Med. 2019;21(5):1049-1057

Costs and effectiveness of whole exome sequencing (WES)

- Preliminary findings suggest WES as first test in diagnostic pathway (instead of the second test) is expected to decrease cost, time to diagnosis, and time in the diagnostic pathway, with minimal change in diagnostic yield



Marshall DA, Degeling K, Tagimacruz T, Seeger TA, Boycott KM, Bernier F, Mendoza-Londona R, MacDonald KV, Hartley T, Hayeems RZ. Costs and effectiveness of whole exome sequencing (WES) in patients with unsolved rare disease through the diagnostic pathway. HTAi 2023 Annual Meeting; Adelaide, Australia: June 24-28, 2023 (oral presentation).

But wait, there's more!
SOLVE is also aiming to…

- Determine the clinical utility of WES, characterized as a change in medical management triggered by WES results
- Determine the pattern and cost of health service utilization from birth to 1 year following WES among patients
  - Will also explore differences between patients who receive a diagnosis, do not receive a diagnosis, or receive an uncertain diagnosis

WATCH THIS SPACE!

Findings from UCAN: Costs in JIA

Rheumatol Ther
https://doi.org/10.1007/s40744-021-00331-x

ORIGINAL RESEARCH

Evaluation of Real-World Healthcare Resource Utilization and Associated Costs in Children with Juvenile Idiopathic Arthritis: A Canadian Retrospective Cohort Study

Luiza R. Grazziotin · Gillian Currie · Marinka Twilt · Maarten J. Ijzerman · Michelle M. A. Kip · Hendrik Koffijberg · Susanne M. Benseler · Joost F. Swart · Sebastiaan J. Vastert · Nico M. Wulffraat · Rae S. M. Yeung · Nicole Johnson · Nadia J. Luca · Paivi M. Miettunen · Heinrike Schmeling · Deborah A. Marshall on behalf of the UCAN CAN-DU, UCAN CURE consortia

EXPERT REVIEW OF PHARMACOECONOMICS & OUTCOMES RESEARCH
https://doi.org/10.1080/14737167.2021.1857241

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ORIGINAL RESEARCH

OPEN ACCESS Check for updates

Costs of medication use among patients with juvenile idiopathic arthritis in the Dutch healthcare system

Michelle M. A. Kip [a,b], Sytze de Roock[b,c,d], Gillian Currie [b,e,f,g], Deborah A. Marshall [b,e,g,h], Luiza R. Grazziotin [b,e], Marinka Twilt [b,g,i], Rae S. M. Yeung [b,j], Susanne M. Benseler [b,g,i], Maud A. Schreijer[a], Sebastiaan J. Vastert[b,c,d,k], Nico Wulffraat [b,c,d,k], Annet van Royen-Kerkhof[b,c,d,k], Joost F. Swart [b,c,d,k] and Maarten J. IJzerman [b,a,l]

RESEARCH ARTICLE Open Acc

Real-world data reveals the complexity of disease modifying anti-rheumatic drug treatment patterns in juvenile idiopathic arthritis: an observational study

Luiza R. Grazziotin[1,2,3,4], Gillian Currie[1,3,4,5], Marinka Twilt[4,6], Maarten J. Ijzerman[7], Michelle M. A. Kip[7], Hendrik Koffijberg[7], Susanne M. Benseler[4,6,8], Joost F. Swart[9,10], Sebastiaan J. Vastert[9,10], Nico M. Wulffraat[9,10], Rae S. M. Yeung[11] and Deborah A. Marshall[1,2,3,4,12*]

RHEUMATOLOGY

Original article

Pharmacological treatment patterns in patients with juvenile idiopathic arthritis in the Netherlands: a real-world data analysis

Michelle M. A. Kip [1,2], Sytze de Roock[2,3], Gillian Currie[4,5,6,7], Deborah A. Marshall [4,6,7], Luiza R. Grazziotin[4], Marinka Twilt[6,8], Rae S. M. Yeung[9], Susanne M. Benseler [6,8], Sebastiaan J. Vastert[2,3,10], Nico Wulffraat[2,3,10], Joost F. Swart [2,3,10,*] and Maarten J. IJzerman[1,11,*]

Arthritis Care & Research
Vol. 74, No. 10, October 2022, pp 1585–1592
DOI 10.1002/acr.24621

American College of Rheumatology
Empowering Rheumatology Professionals

Costs of Hospital-Associated Care for Patients With Juvenile Idiopathic Arthritis in the Dutch Health Care System

Michelle M. A. Kip,[1] Sytze de Roock,[2] Inge van den Berg,[3] Gillian Currie,[4] Deborah A. Marshall,[4] Luiza R. Grazziotin,[4] Marinka Twilt,[4] Rae S. M. Yeung,[5] Susanne M. Benseler,[4] Sebastiaan J. Vastert,[2] Nico Wulffraat,[2] Joost F. Swart,[2] and Maarten J. IJzerman[6]

Findings from UCAN: Quality of Life JIA

Grazziotin et al. Pediatric Rheumatology (2022) 20:51
https://doi.org/10.1186/s12969-022-00713-7

Pediatric Rheumat

RESEARCH ARTICLE Open Ac

Factors associated with care- and health-related quality of life of caregivers of children with juvenile idiopathic arthritis

Luiza R. Grazziotin[1,2,3,4], Gillian Currie[1,3,4,5], Marinka Twilt[2,4,6], Maarten J. IJzerman[7], Michelle M. A. Kip[7], Hendrik Koffijberg[7], Gouke Bonsel[8], Susanne M. Benseler[4,6,9], Joost F. Swart[10,11], Sebastiaan J. Vastert[10,11], Nico M. Wulffraat[10,11], Rae S. M. Yeung[12], Wineke Armbrust[13], J. Merlijn van den Berg[14] and Deborah A. Marshall[1,2,3,4*]

Pediatric Rheumatology 2022, 20(Suppl 2):75
https://doi.org/10.1186/s12969-022-00729-z

Pediatric Rheumatology

MEETING ABSTRACTS Open Access

Proceedings of the 28th European Paediatric Rheumatology Congress (PReS 2022)

Prague, Czech Republic. 20-23 September 2022

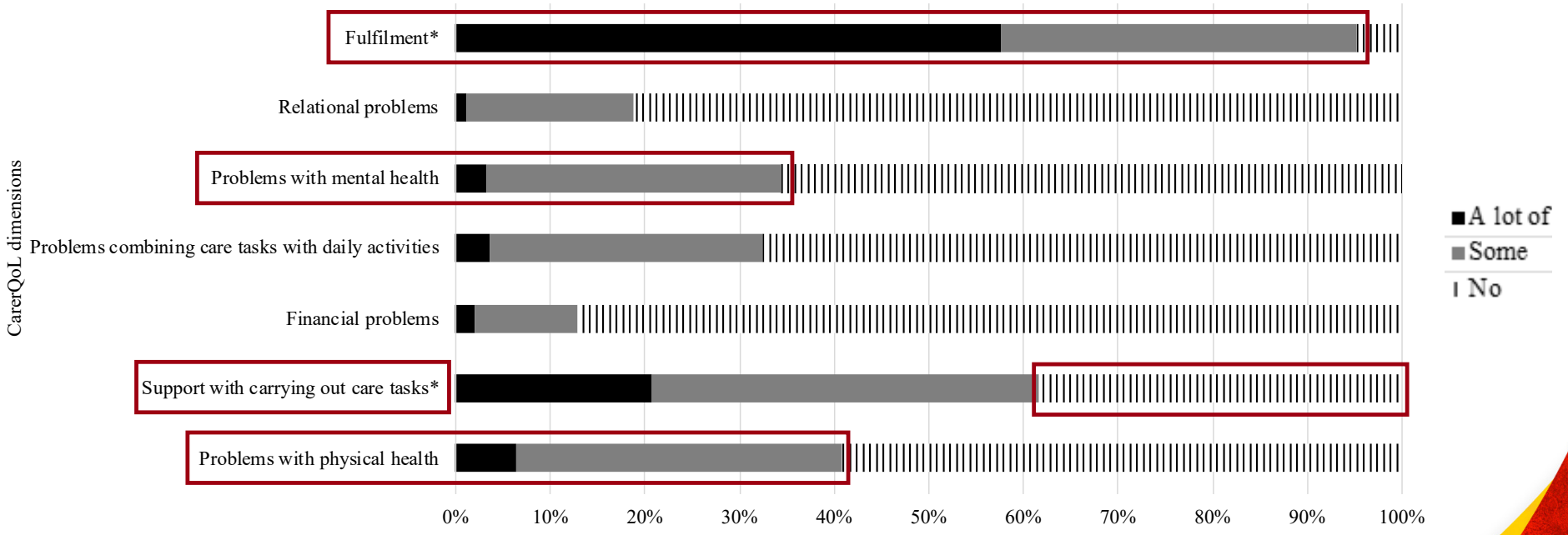
Published: 7 September 2022

P420.

Present and accounted for: the workplace productivity loss for parents of children with juvenile idiopathic arthritis

L. Grazziotin[1], G. R. Currie[1], S. Cantarutti[1], S. M. Benseler[1], J. F. Swart[2], M. M. Kip[3], M. J. IJzerman[4], M. Twilt[1], S. J. Vastert[2], N. M. Wulffraat[2], R. S. Yeung[5], D. A. Marshall[1] on behalf of on behalf of UCAN CAN-DU and UCAN Cure Consortium

Caregiving Quality of Life in Parents



- Grazziotin et al. Factors Associated with Care- and Health-Related Quality of Life of Caregivers of Children with Juvenile Idiopathic Arthritis. Pediatric Rheumatology 2022;20:51

Productivity Loss Among Parents

- Preliminary fundings
- 12% of parents had made changes in their work commitment due to their child's JIA

73% reduced working hours

13% stopped working altogether

- For those working, overall work impairment = 26%
- Mean impairment to usual activities: 20%

PAVE: Producing and Arthritis Value Framework with Economic Evidence – Paving the Way for Rare Childhood Diseases

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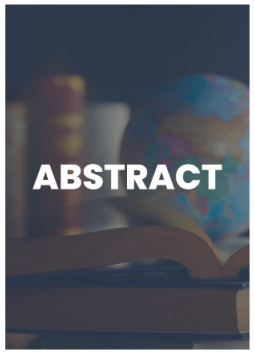
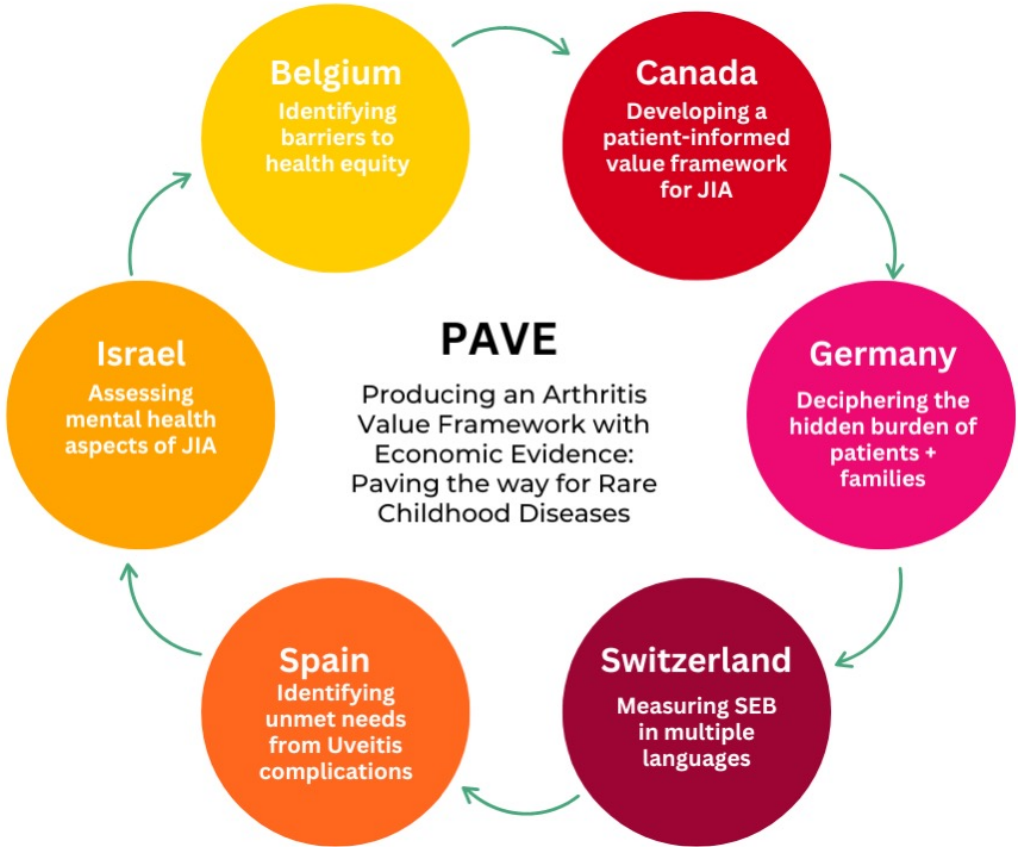
EUROPEAN JOINT PROGRAMME RARE DISEASES

Joint Transnational Call 2021 (JTC2021)

PAVE: Producing an Arthritis Value-Framework with Economic Evidence – Paving the Way for Rare Childhood Diseases



Arthritis in children and youth is a group of rare and ultra-rare diseases. Children of all ages suffer from joint pain, stiffness and swelling slowly destroying they joints and resulting in life-long disability. Pain, decreased mobility and severe fatigue dramatically impact the child's everyday quality of life and the life of the entire family. The burden of arthritis is far larger than medication costs. Children suffer – miss school days, play dates, and other childhood milestones. They often have poor mental health, struggle with education and look at a future of low paid jobs or unemployment due to disability. Families suffer – loss of employment, full time, long-term care provision to their child and home tutoring, making up for the many educational demands. There is a dramatic hidden burden of arthritis. The PAVE team is a strong international partnership of 6 countries including clinical leaders, researchers, social scientists and strong engagement with patient organizations. Our team will focus on the unmet needs of children and families and empower them with new ways to address the hidden burden of arthritis. The PAVE team will build a first-of-its kind value framework that will unite the voices of children and families with data-informed calls to action to measure and ultimately reduce the cost and burden of childhood arthritis. Together, we will improve the lives of children living with arthritis, reduce inequities and pave the way for improved lives for people of all ages living with rare diseases. Children with arthritis and their families need a voice.



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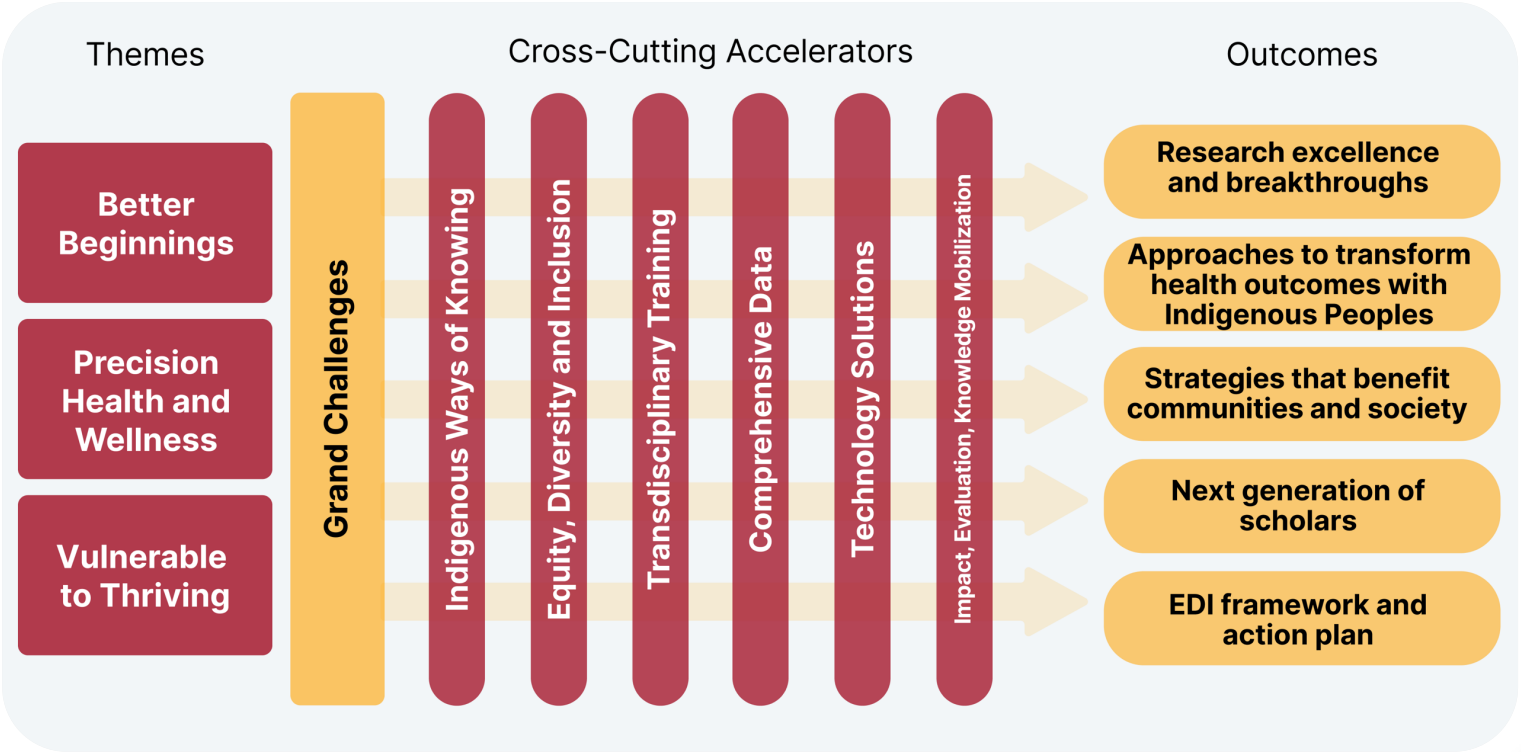
one child every child

A Transformational Child Health Research Initiative



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CANADA FIRST RESEARCH EXCELLENCE FUND

Moving Forward…fully capturing SE impact

COSTS

DIAGNOSTIC ODYSSEY COSTS

OUTCOMES

QUALITY OF LIFE

- Refining and further developing the cost framework
  - Beyond health care costs
- Better measurement of health care system costs
  - Limitations of administrative data
  - Role of registries
- Need to incorporate the diagnostic odyssey
- Going beyond costs:
  - Family Impacts
  - Quality of Life (patient & caregiver)
  - Patient Preferences and Input

If you can't measure it, you can't manage it.

- Peter Drucker

- If what gets measured gets improved, it is imperative that we measure the socioeconomic burden of rare disease.
- To support resource allocation for rare diseases, understand needs of patients and families, inform planning of health care and other services and support:
  - We need the data – and we need to measure!

Thank you!

Contact:
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Deborah Marshall damarsha@ucalgary.ca

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https://cumming.ucalgary.ca/research/health-economics/home

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